Loss of Heterozygosity

## Find the pertinent genes and likely diagnosis in seconds

| Pertinent positive findings  |  |  |             |  |  |  |
|--|--|--|-------------|--|--|--|
| Onsets can be at an age, by an age, or unknown   |  |  |             |  |  |  |
| Req'd  | Onset  | Finding  | Pertinence  |  |  |  |
| nequ   | ≤1m  | Nystagmus, non-rotary  | T et unence |  |  |  |
|  | ≤6m  | Hyperreflexia  |             |  |  |  |
|  | @1m  | Microcephaly   |             |  |  |  |
|  | 1  | CT or MRI: brainstem atrophy or hypoplasia   | high→       |  |  |  |
| Pertin   | ent nega   | ative findings   |             |  |  |  |
|  | Absent   | Finding  | Pertinence  |  |  |  |
|  | х  | CT or MRI: corpus callosum hypogenesis   |             |  |  |  |
|  | х  | TSEN54 gene mutations (biallelic)  |             |  |  |  |
|  | x  | CT or MRI: cerebral cortex atrophy or hypoplasia   |             |  |  |  |
|  | x<br>x   | X-ray or CT: brain calcifications<br>Creatine kinase high  |             |  |  |  |
|  | x  | Regression   |             |  |  |  |
|  |  |  | high-       |  |  |  |
|  |  |  |             |  |  |  |
| Family   | / history  | 7  |             |  |  |  |
|  |  | 7<br>on known clinical findings  |             |  |  |  |
| Family hi<br>1 of 2 br<br>Mother   | story based<br>others affe<br>not affecte  | on known clinical findings<br>ected<br>d   |             |  |  |  |
| 1 of 2 br<br>Mother<br>Father r  | story based<br>others affe   | on known clinical findings<br>eted<br>d<br>l   |             |  |  |  |
| Family hi<br>1 of 2 br<br>Mother<br>Father r<br>Consang  | story based<br>others affe<br>not affected<br>ot affected<br>guinity: 1st  | on known clinical findings<br>eted<br>d<br>l   |             |  |  |  |
| Family hi<br>1 of 2 br<br>Mother<br>Father r<br>Consang  | story based<br>others affe<br>not affected<br>ot affected<br>guinity: 1st<br><b>cential</b>  | on known clinical findings<br>exted<br>d<br>cousin   | Probability |  |  |  |
| Family hi<br>1 of 2 br<br>Mother<br>Father r<br>Consang<br>Differ<br>Diseas  | story based<br>others affecter<br>not affecter<br>ot affecter<br>guinity: 1st<br><b>cential</b><br>se  | on known clinical findings<br>exted<br>d<br>cousin   | Probability |  |  |  |
| Family hi<br>1 of 2 br<br>Mother r<br>Father r<br>Consang<br>Differ<br>Diseas<br>LIS2: RI  | story based<br>others affe<br>not affected<br>out affected<br>guinity: 1st<br>rential<br>se<br>ELN-relate  | on known clinical findings<br>cted<br>d<br>cousin<br>diagnosis   | Probability |  |  |  |
| Family hi<br>1 of 2 br<br>Mother r<br>Father r<br>Consang<br>Differ<br>Diseas<br>LIS2: RI<br>PCH8: 1   | story based<br>others affe<br>not affected<br>out affected<br>guinity: 1st<br>rential<br>se<br>ELN-relate  | on known clinical findings<br>eted<br>d<br>cousin<br><b>diagnosis</b><br>wl lissencephaly. AR<br>ellar hypoplasia. CHMP1A-related  | Probability |  |  |  |
| Family hi<br>1 of 2 br<br>Mother<br>Father r<br>Consang<br>Differ<br>Diseas<br>LIS2: RI<br>PCH8: J<br>CDG1A:   | story based<br>others affe<br>not affecte<br>out affecte<br>guinity: 1st<br>rential<br>se<br>ELN-relate<br>pontocereb<br>PMM2-re   | on known clinical findings<br>eted<br>d<br>cousin<br><b>diagnosis</b><br>wl lissencephaly. AR<br>ellar hypoplasia. CHMP1A-related  | Probability |  |  |  |
| Family hi<br>1 of 2 br<br>Mother<br>Father r<br>Consang<br>Differ<br>Diseas<br>LIS2: RI<br>PCH8: [<br>CDG1A:<br>PCH10:   | story based<br>others affe<br>not affecte<br>out affecte<br>guinity: 1st<br>rential<br>se<br>ELN-relate<br>pontocereb<br>PMM2-re<br>Pontocere  | on known clinical findings<br>cted<br>d<br>cousin<br><b>diagnosis</b><br>dlissencephaly, AR<br>ellar hypopolasia, CHMP1A-related<br>lated  | Probability |  |  |  |
| Family hi<br>1 of 2 br<br>Mother Father r<br>Consang<br>Differ<br>Diseas<br>LIS2: RI<br>PCH8: [<br>CDG1A:<br>PCH10:<br>VLDLR-  | story based<br>others affe<br>not affecte<br>out affecte<br>out affecte<br>out affecte<br>unity: 1st<br>eential<br>se<br>ELN-related<br>pontocerel<br>PMM2-re<br>Pontocere<br>related ce   | on known clinical findings<br>cted<br>d<br>cousin<br>diagnosis<br>dissencephaly, AR<br>eilar hypoplasia. CHMP1A-related<br>lated<br>bellar hypoplasia. CHP1-related  | Probability |  |  |  |
| Family hi<br>1 of 2 br<br>Mother F<br>Father r<br>Consang<br>Differ<br>Diseas<br>LIS2: RI<br>PCH8: [<br>CDG1A:<br>PCH10:<br>VLDLR-<br>LISX1: 1   | story based<br>others affecte<br>out | on known clinical findings<br>ceted<br>d<br>i<br>cousin<br><b>diagnosis</b><br>dlissencephaly, AR<br>ellar hypoplasia, CHMP1A-related<br>lated<br>bellar hypoplasia, CLP1-related<br>rebellar hypoplasia   | Probability |  |  |  |
| Family hi<br>1 of 2 br<br>Mother r<br>Father r<br>Consang<br>Differ<br>Diseas<br>LIS2: RI<br>PCH8: I<br>CDG1A:<br>PCH10:<br>VLDLR-<br>LISX1: I<br>PCH1B:   | story based<br>others affe<br>not affecte<br>guinity: 1st<br>rential<br>se<br>ELN-relate<br>contocereb<br>PMM2-re<br>Pontocere<br>related ce<br>DCX-relate<br>pontocere  | on known clinical findings<br>ceted<br>d<br>i cousin<br>diagnosis<br>dlissencephaly, AR<br>ellar hypoplasia, CHMP1A-related<br>lated<br>bellar hypoplasia, CLP1-related<br>rebellar hypoplasia<br>dissencephaly, X-linked<br>bellar hypoplasia<br>dissencephaly, X-linked  | Probability |  |  |  |
| Family hi<br>1 of 2 br<br>Mother<br>Father r<br>Disear<br>Disear<br>LIS2: RI<br>PCH8: p<br>CDG1A:<br>PCH10:<br>VLDLR-<br>LISX: 1<br>PCH1B:<br>PCH2: p  | story based<br>others affe<br>not affected<br>out affected<br>guinity: 1st<br><b>rential</b><br>se<br>ELN-relate<br><u>sontocereb</u><br>PMM2-re<br>Pontocere<br>related ce<br>DCX-relate<br>pontocereb  | on known clinical findings<br>cted<br>d<br>i<br>cousin<br><b>diagnosis</b><br>dlissencephaly, AR<br>ellar hypoplasia. CHMP1A-related<br>lated<br>bellar hypoplasia. ACHP1-related<br>ebellar hypoplasia. RCMP2-related<br>ebellar hypoplasia. RCMP2-related<br>ebellar hypoplasia.   | Probability |  |  |  |
| Family hi<br>1 of 2 br<br>Mother<br>Father r<br>Consang<br>Differ<br>Diseas<br>LIS2: RI<br>PCH8: [<br>PCH8: [<br>PCH10:<br>VLDLR-<br>LISX1: I<br>PCH1B:<br>PCH2: [<br>Aicardi-   | story based<br>others affe<br>not affected<br>guinity: 1st<br><b>rential</b><br>se<br>ELN-related<br>yontocereb<br>PMM2-re<br>Pontoceree<br>related ce<br>OCX-related<br>pontocereb<br>Goutières   | on known clinical findings<br>cted<br>d<br>t cousin<br>diagnosis<br>dlissencephaly, AR<br>ellar hypoplasia, CHP1-related<br>lated<br>bellar hypoplasia, CHP1-related<br>chellar hypoplasia, CHP1-related<br>chellar hypoplasia, CHP1-related<br>dlissencephaly, X-linked<br>dlissencephaly, X-linked<br>signature of the second se | Probability |  |  |  |
| Family hi<br>1 of 2 br<br>Mother r<br>Consang<br>Differ<br>Diseas<br>LIS2: RI<br>PCH8: [<br>CDG1A:<br>PCH10:<br>VLDLR-<br>LISX1: I<br>PCH18:<br>PCH2: [<br>Aicardi-<br>PCH14:<br>PCH14:  | story based<br>others affected<br>out affected<br>guinity: 1st<br><b>cential</b><br>se<br>ELN-related<br>contocereb<br>PMM2-re<br>Pontocere<br>related ce<br>DCX-related<br>pontocereb<br>Goutières<br>pontocereb  | on known clinical findings<br>cted<br>d<br>cousin<br>diagnosis<br>dlissencephaly. AR<br>ellar hypoplasia. CHMP1A-related<br>lated<br>bellar hypoplasia. CLP1-related<br>rebellar hypoplasia<br>dlissencephaly. X-linked<br>bellar hypoplasia 2<br>syndrome. AR<br>bellar hypoplasia 2<br>syndrome. AR<br>bellar hypoplasia 2<br>syndrome. AR   | Probability |  |  |  |
| 1 of 2 br<br>Mother Tehter 1<br>Consang<br>Differ<br>Diseau<br>LIS2: RI<br>PCH8: p<br>CCDG1A:<br>PCH0:<br>VLDLR-<br>LISXI: 1<br>PCH1B:<br>PCH2: p<br>CH2: p<br>Aicardi-<br>PCH1A:<br>MASA: 1   | story based<br>others affected<br>out affected<br>guinity: 1st<br><b>cential</b><br>se<br>ELN-relate<br>contocereb<br>PMM2-re<br>Pontocered<br>pontocered<br>pontocered<br>Goutières.<br>pontocered<br>Goutières.  | on known clinical findings<br>ceted<br>d<br>cousin<br>diagnosis<br>elliar hypoplasia. CHMP1A-related<br>lated<br>bellar hypoplasia. CLP1-related<br>bellar hypoplasia. EXOSC3-related<br>elliar hypoplasia. EXOSC3-related<br>elliar hypoplasia. 2<br>syndrome. AR<br>bellar hypoplasia. 2<br>syndrome. AR<br>bellar hypoplasia. Shuffling. adducted thumbs  | Probability |  |  |  |
| Family hi<br>1 of 2 br<br>Mother<br>Father r<br>Consang<br>Differ<br>Diseas<br>LIS2: RJ<br>PCH8: p<br>CCG1A:<br>PCH0: p<br>CCG1A:<br>CCG1A:<br>PCH0: p<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CCG1A:<br>CC | story based<br>others affe<br>not affected<br>yunity: 1st<br>ential<br>se<br>ELN-relate<br>sontocereb<br>PMM2-re<br>PMM2-re<br>PMM2-re<br>PMM2-re<br>related ce<br>DCX-relate<br>pontocere<br>goontocereb<br>Goutières<br>pontocere<br>mental ret<br>elizaeus-N  | on known clinical findings<br>cted<br>d<br>cousin<br>diagnosis<br>dlissencephaly. AR<br>ellar hypoplasia. CHMP1A-related<br>lated<br>bellar hypoplasia. CLP1-related<br>rebellar hypoplasia<br>dlissencephaly. X-linked<br>bellar hypoplasia 2<br>syndrome. AR<br>bellar hypoplasia 2<br>syndrome. AR<br>bellar hypoplasia 2<br>syndrome. AR   | Probability |  |  |  |

# **Key Benefits**

#### Focused on your patient.

Uses your patient's pertinent positive and negative findings to generate a list of candidate genes in order of pertinence

Fast. Analysis in seconds, enables useful discussion with referring physician, when needed

Accurate. Reduces errors and inconsistencies

### **Key Features**

- Automatically correlates the list of patient findings with the genes in chromosomal regions
- Identifies and ranks the genes by pertinence for this patient
- Enables a quick call to the ordering physician to focus on additional findings that help reach a conclusion, relying on the "useful findings" and "useful tests" features

### **CONSULT** on genes in regions of loss of heterozygosity

| VLDLR-related cerebe<br>JBTS25: Joubert synd  | Portinent gene avgesities                 |                 |                          |  |
|---|---|-----------------|--------------------------|--|
| Homocystinuria, mega<br>Hartnup disease, symp | 5 V V VLDLK gene variants (blanenc)       |                 |                          |  |
| MRT5: mental retarda                          |   |                 |                          |  |
| Adrenoleukodystrophy                          |   | Differen        | tial Dx                  |  |
| LIS2: RELN-related li                         |   | Incidenta       | l genes                  |  |
| Retinal cone dystrophy                        |   | Discover        | v genes                  |  |
| Multiple sulfatase defi                       |   |                 |                          |  |
| PCH8: pontocerebella                          |   | Panel of        | genes                    |  |
| Zellweger syndrome                            | 5 • V • NPHP4 gene variants (biallelic)   | Loss of h       | s of heteroz<br>rognosis |  |
| CDG1A: PMM2-relate                            | 5 V V GLDC gene variants (biallelic)      | Progn           |                          |  |
| PCH10: Pontocerebell                          | 5 V V ESPN gene variants (biallelic)      | Assess f        | inding                   |  |
| LISX1: DCX-related li                         | SLCIAI gene variants (biallelic)          |                 |                          |  |
| PCH1B: pontocerebell                          | 5 V V V CKBN gene variants (bianenc)      | Profile finding |                          |  |
| PCH2: pontocerebella                          | 5 • V • SLC6A3 gene variants (biallelic)  | Database        |                          |  |
| Dubowitz syndrome                             | 5 🔽 🗸 💌 B3GALT6 gene variants (biallelic) | Search          | File                     |  |
| Aicardi-Goutières syn                         | 5 V V IKNII gene variants (bianenc)       | Start           | Help                     |  |
| Senior-Løken syndrom                          | S V V GLISS gene variants (blanenc)       |                 |                          |  |
| PCH1A: pontocerebell                          | 5 V V PLEKHG5 gene variants (bianenc)     |                 |                          |  |
| Multiple sulfatase defi                       |   |                 |                          |  |
|   | More tips                                 | Summary         | Note                     |  |

To get a license, email:

SimulConsult.com